SOME CONGENITAL DEFORMITIES OF THE RECTUM, ANUS, VAGINA AND URETHRA

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by
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Classification of these deformities

There are four main groups of factors which account for abnormalities in the new-born :

(1) Those due to genetic variations.

(2) Those due to mechanical variations in the relations between the fœtus and its surroundings.

(3) Those due to toxic influences, such as rubella or lack of vitamins.

(4) Those due to injuries during the act of birth.

It is the first group alone that is considered in this paper. The deformities composing it can be classified according to the various processes that have gone wrong in the strangely complicated and inexplicable manner in which the human body forms itself. These may be grouped as follows :—

(1) *Failures of formation*, when some part of the body does not develop, in whole or in part.

(2) *Extra formations*, when a structure is present at birth in addition to those of the normal body.

(3) *Wrong formations*, when a structure is mal-formed.

(4) *Incomplete migration*, when a structure that is developed in one part of the body, but should take up a permanent position in another, fails to complete the journey.

(5) *Erratic migration*, when a migrating structure leaves the correct path.

(6) *Incomplete fusion*, when the process of the joining of two structures that is necessary for the completion of a part of the body fails entirely or in part.

(7) *Excessive fusion*, when the process of junction proceeds further than it should.

(8) *Failures of atrophy*, when some part of the fœtus that should disappear in the process of development is present at birth, in whole or in part.

The commoner types of deformity in the parts concerned will be discussed in terms of these eight categories.

DEFORMITIES OF THE RECTUM AND ANUS

In discussing this subject one is handicapped by the lack of a generally accepted account of the development of this region. The heretical hypothesis of Wood Jones that the rectum develops as an offshoot from

the hind-gut, rather than the orthodox one that it is formed by the division of the cloaca by the growth of a septum, best explains the stock deformities of the region. I think the surgeon has a right to demand from the anatomist that the hypotheses of embryology should accord with the undoubted facts of clinical experience.

There is also the difficulty that the problem of the closure of the anus, like the not very dissimilar one of the closure of the nasopharynx, has slipped unnoticed through the curious gap that is traditional in medicine between the anatomists who study the structure of the body, and the physiologists who study its working. There is no authoritative answer, for instance, to the simple and important question of whether the levatores ani alone can give satisfactory control.

The discussion of these questions of development and function is, however, a matter for another paper, which I hope to publish before long in collaboration with Mr. Douglas Stephens, to whom I am indebted for much stimulation and help, particularly in the understanding of the " covered anus " and " covered vagina "

FAILURES OF FORMATION

Rectal agenesis. This is one of the many conditions lumped together under the term of " imperforate anus " with or without " fistulæ." I think that much bad treatment in this region is due to misleading terminology and that a determined attempt should be made to get a more accurate one.

In these comparatively rare cases the bowel ends in a completely blind sac, a greater or less distance above the normal site of the anus. There has been no sphincter mechanism in this blind end in any case that I have seen. I know that certain cases are described in which there has been merely a thin diaphragm over the anus, incision of which has produced a normally working opening ; but I think there can be no doubt that many, if not all, of these were examples of the microscopic or covered anus which is described later.

At birth such blind endings are widely distended with gas and viscous meconium, and their walls are accordingly very thin. This makes them almost impossible to handle surgically, and the babies who have them die fairly rapidly from distension if unrelieved. In this they differ from those cases which have a vent, however small, for gas and meconium into the urethra or vagina. In these, life may persist without operation for surprisingly long periods ; or even indefinitely, in those with the larger and lower openings.

Diagnosis. This condition must be distinguished from a recto-urethral fistula in planning the long-term treatment, and from a " microscopic anus " or " covered anus " as regards immediate operation. A careful watch should be kept on the tip of the penis for the emergence of minute quantities of meconium and repeated examinations of the perineum made to detect tiny specks of the same material there. X-rays should

be taken, after the baby has been held in the inverted position for five minutes to allow the sticky meconium to drain from the blind end of the bowel and to be replaced by a bubble of air ; while the normal site of the anus is marked by a small piece of lead.

Treatment. If there is marked bulging in the normal anal situation on crying, it is justifiable to make a very small incision and to carry it to a depth of a quarter of an inch to exclude what must be the very uncommon condition of a rectal diaphragm. The traditional dissection upwards through the pelvis is, in my opinion, a disastrous mistake which should never be made. In the minute perineum and pelvis of a new-born infant it is impossible to do this without wrecking the entire pelvic floor, on whose integrity continence after operation must depend ; and, if the bowel is found, it is impossible to free it enough to bring it down to the skin without tension that inevitably results in subsequent retraction. The result is apt to be an unsatisfactory colostomy in an uncontrollable place, with bouts of obstruction due to contraction of scar tissue round the opening, alternating with bouts of incontinence when this opening is dilated.

Treatment should be considered from two points of view; the primary saving of the baby's life, and the secondary securing (if possible) of a normally working anus. A colostomy to allow the bowel contents to escape should be done in these cases without delay, but there are two most important requirements for this ; it should not hinder any subsequent approach to the critical region of the pelvis, and it should be capable of safe and easy closure. The only opening that satisfies these conditions is one through the middle of the transverse colon, made with a carefully constructed deep spur that can be crushed when closure is desired. Anyone working on these conditions knows only too well, however, that he almost invariably has to contend with a fæcal fistula or a large prolapse low in the left inguinal region.

For the secondary operation no proved satisfactory technique has yet been worked out, and I am inclined to think that there are some cases with a high blind end ending to the gut in which the provision of a working perineal anus is surgically impossible. This pessimism is reinforced by the co-existence in many of these children of gross defects of development of the sacrum and coccyx.

There are four routes for reaching the inaccessible region concerned. The first and most common is the dissection up through the perineum which has already been condemned. The second is that through the abdomen, which is apt to become more difficult the more favourable the case is ; that is to say, the deeper the blind end of the gut lies in the pelvis. The third is through an incision between the coccyx and the sacrum, letting the surgeon open up the loose areolar tissue between the uninjured levatores ani and the peritoneum. With the aid of a frame retractor that will pull in many directions at once, and of a brilliant head-light that will alone enable the surgeon to see into such

deep and narrow clefts, the freeing and manipulation of the blind end of gut is less difficult than might be imagined. It can, when sufficiently freed, be brought down through a small opening, stretched, not cut, in the floor of the pelvis, and there sutured without tension. It is too early yet to say how such an opening will work, but there is reason for restrained optimism so far. (Fig. 1.)

The fourth approach is only feasible when a urethral fistula is present. It consists in splitting up the urethra in the perineal region and carrying the cut back until the fistula is reached, in the knowledge that the urethra can be closed with certainty when desired. The opening into the gut is kept dilated until its tendency to contract has ceased. I have tried this method on two cases, but it is too early yet to evaluate it.

Stenosis of the anus. This is an obvious condition, although some of its anatomical peculiarities are puzzling, especially the frequent occurrence of a thick band of tissue running anterio-posteriorly across the anal site, with a minute opening on one side of it. Occasionally, this band is completely detached in its centre. (Figs. 2, 3 and 4.)

Treatment is the obvious and satisfactory one of dilatation, which I think is best performed without any incision, although an anæsthetic will be necessary for the first occasion. The bands of tissue should be simply cut away, and no attempt made to suture the raw surfaces thus produced ; they will heal over quickly and well.

Microscopic anus. In a fair number of cases in careful watch of what appears to be an intact perineum there will be observed to appear a small spot of meconium, no larger than a fly speck, emerging from an opening so small as to be invisible except under a lens. (Fig. 5.)

Treatment. The blunt end of a surgical needle makes a suitable fine probe for the first investigation of such an opening. Instruments of increasing size are passed in succession, and a few months of dilatation will produce a normally working anus.

FAILURE OF ATROPHY

Recto-urethral fistula. It is common for a communication to remain between the gut and urinary system—which should have become shut off from each other—in the form of a small passage between the lower end of a mal-developed bowel and the prostatic region of the urethra. It is curious that this fistula is never found with a properly developed rectum, but that, if it is present, it forms the lowest part of the intestinal tube. The great majority of the cases described as " imperforate anus " in the male are of this type ; it is comparatively rare for the bowel to have a completely blind end. These openings are always necessarily small, because of the calibre of the urethra, and have no sphincter mechanism.

Treatment. This is the same as in the simple rectal agenesis that has been described, except for the treatment of the fistula at operation. This can be isolated and divided between curved artery forceps, the one next to the urethra being oversewn with a running fine catgut stitch before

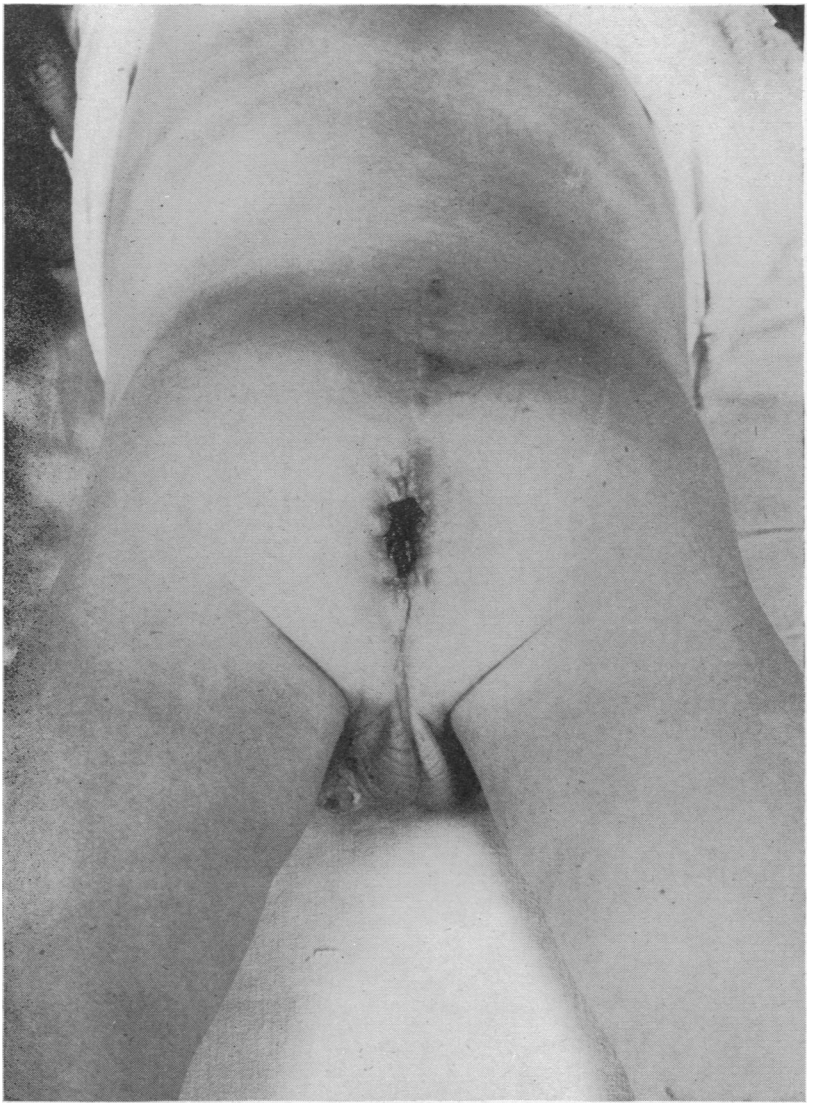

Fig. 1. An agenesis of the rectum after operation. The scar of the approach between the coccyx and the sacrum can be seen. The bowel mucosa is showing on the surface. Control is promising in this case.

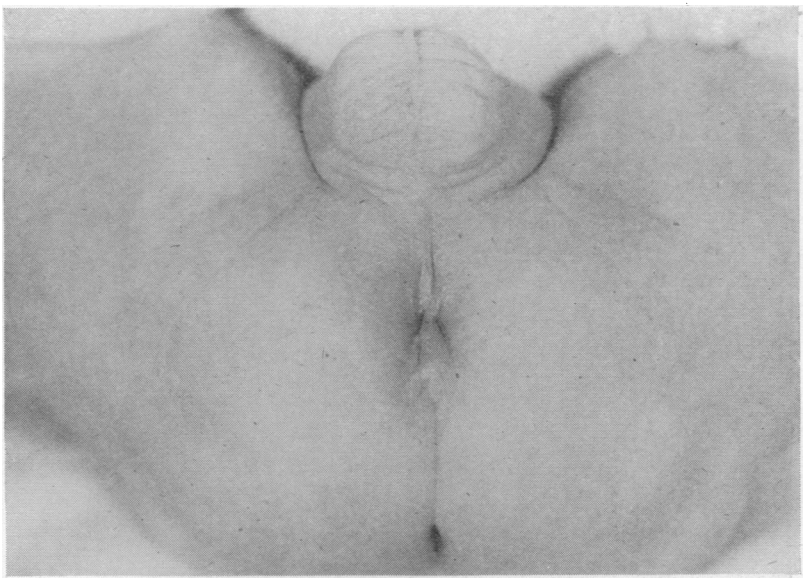

Fig. 2. Stenosis of the anus. The minute opening was to the right side of the longitudinal bar. The bar was excised and the opening dilated with excellent results.

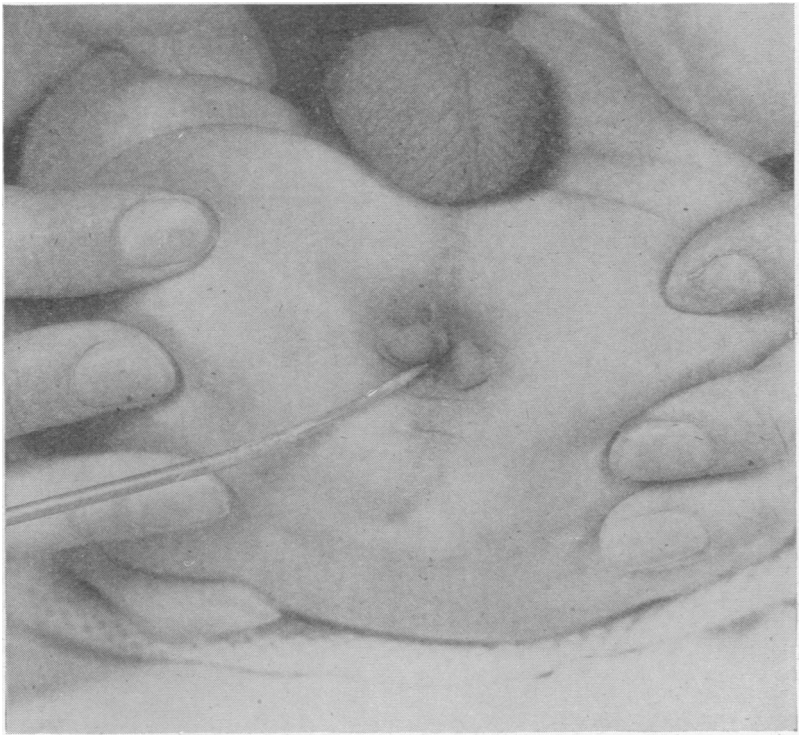

Fig. 3. Stenosis of the anus. The probe points to the minute opening. Excision of the fleshy tags and dilatation gave excellent results.

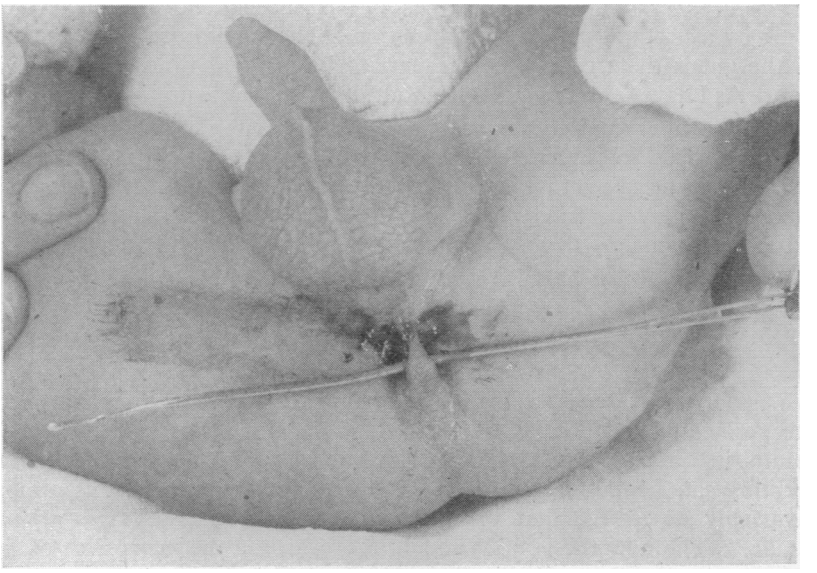

Fig. 4. Showing a completely detached longitudinal band over a stenosed anus.

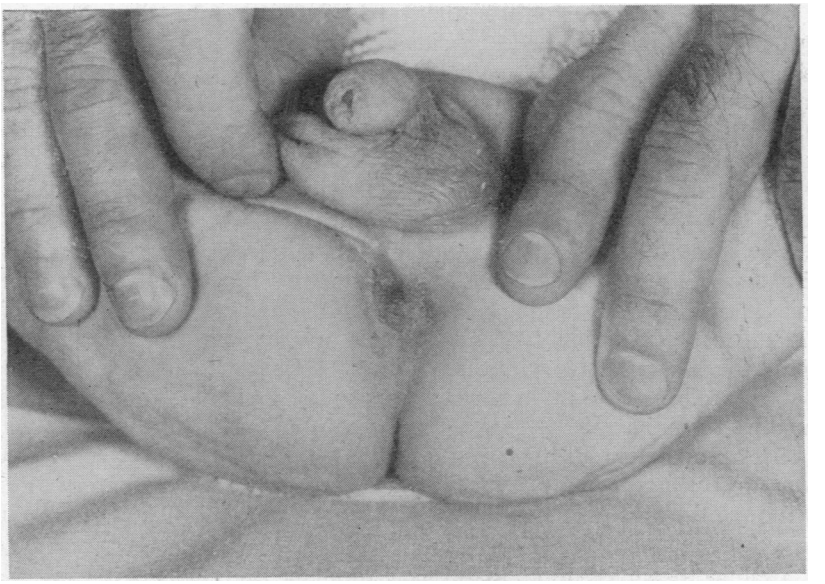

Fig. 5. Apparently imperforate anus which had a microscopic opening, dilatation of which gave excellent results.

the forceps are removed. The bladder is subsequently drained for a week by a urethral catheter with suction. The small opening in the end of the gut is then, of course, the part to be brought through the pelvic floor. As I have said, I am unsure about the value of the urethral approach.

High recto-vaginal fistula. In certain, fortunately rare, cases there is a narrow fistula between the lower end of the gut and the vagina, at a point corresponding to the level of the prostatic urethra in the male. Treatment may be tried along the same lines as in the recto-urethral fistula in the male. In the examples of this condition which I have seen, there have, however, always been gross accompanying defects of the female genitalia.

ERRATIC MIGRATION

Vaginal ectopic anus. This is the commonest of all mal-formation of the perineum, and the easiest to treat. It is also one which frequently results in an unnecessarily ruined life, and a good deal of the blame for this can be put upon the misleading name by which it is almost invariably described, that of "imperforate anus with a recto-vaginal fistula." The hypothesis of Wood Jones regarding the outgrowth of the rectum as a bed from the hind-gut, explains the fact that this outgrowth not infrequently misses the proctodeal pit where it should form a normal anus, and ends up further forward than it should.

Its mildest degree is what I call the—

Shot-gun perineum. In this rather rare deformity a normally functioning anus and vagina open side by side without the intervention of a strip of skin which should normally separate them. (Fig. 6.) There is no interference with function, and women in this condition may marry and have children without ever suspecting their slight departure from the normal. Whether any surgeon has ever tried the difficult and risky operation that would be necessary to separate the two orifices in such cases I do not know, but I hope not.

The importance of this condition is that if from severer degrees of this deformity we can gain as a final result this functionally satisfactory minor degree, we should be content with that, refusing dangerous risks for the possibility of an unnecessary anatomical perfection. I think the analogy holds with the treatment of hypospadias. In the mildest degree of this deformity, consisting merely of a slight divergence of the meatus from its exactly normal position, there is no interference with structure or function sufficient to justify operation. In the severe degrees the surgeon accordingly may be satisfied to produce such a formation, although the meatus may not be exactly in its normal position at the very tip of the glans.

It must be understood that this is quite a different condition from recto-urethral fistula in the male, or the high recto-vaginal opening in the female. An ectopic anus is a true anus, complete with normal nerve

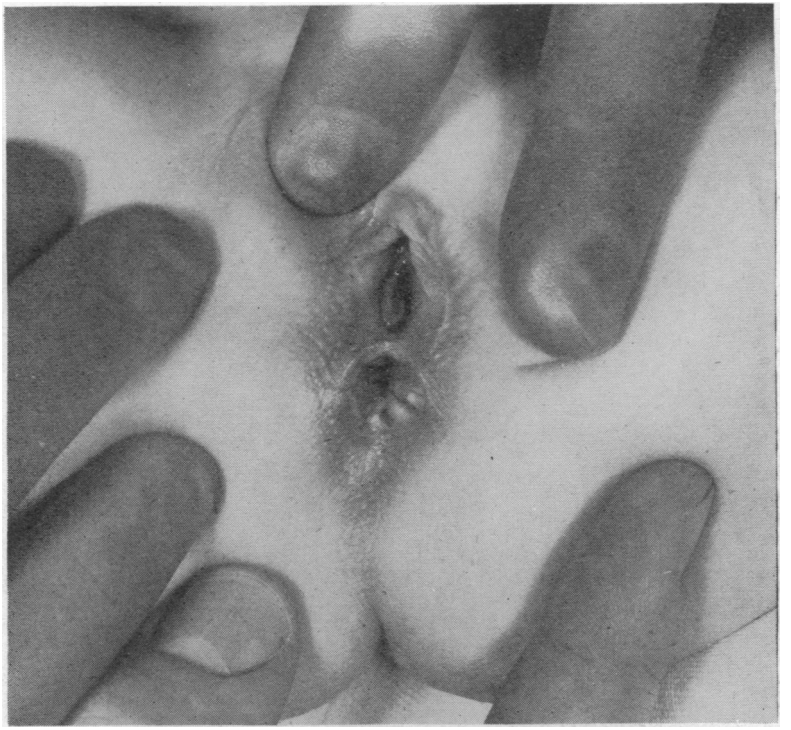

Fig. 6. Congenital " shot-gun perineum," anus and vagina opening side by side.

control for opening and closing. The most common situation for a vaginal anus is fortunately the easiest of all to treat; it is just inside the vaginal opening, readily visible on examination by a suitable speculum.

Treatment. In the great majority of cases the surgeon is at present led astray by the suggestions of the name of " imperforate anus with recto-vaginal fistula " which I have already criticised. He feels the absent anus must be constructed and the fistula closed. Accordingly, he makes an opening into the bowel through the normal site of the anus, and ignores or tries to close the true anus lying in front of this. The result is continual leakage through both openings and complete misery for the child.

There is another method of operating which has a delusive attractiveness about it when illustrated in a text book. It consists in transplanting the displaced anus bodily backwards after detaching it from the vagina. I cannot say that this proceeding is never successful, although I believe success to be only possible in the slightest degrees of the deformity. The possible gains by it seem to me to be in no way balanced by the extreme likelihood of irremediable disaster. If one tries to draw this

operation as it appears from the sagittal plane, instead of from the surface of the perineum, the reason for these disasters becomes clear. There is a large open space left in front of the lower part of the rectum, which must inevitably become septic and which can in no way be filled. The anal opening is extremely flimsy and cannot be sutured in position firmly enough to resist the pull of this granulating area in front. The common result is that the anus becomes inextricably buried in a dense mass of scar tissue, and a permanent colostomy is the only resource.

The operation which I recommend is a simple backward incision from the displaced opening right across the normal situation of the anus, made by placing one blade of a pair of dissecting scissors in the bowel while the other lies on the skin. This opening is then kept dilated by the passage of a lubricated metal Hegars sound, size about number eight. No attempt should be made to suture the raw surfaces thus produced, and after a month or two they will be covered with supple and satisfactory new skin.

A perineum produced in this way goes on improving in appearance and function as growth proceeds, and ends by being indistinguishable in most cases from the congenital shot-gun perineum. If a surgeon should persist in trying to construct an absolutely normal perineum, this is surely an indispensable preliminary to it.

In the frequent cases in which an opening in the normal situation has been surgically made the child will be greatly relieved if one blade of a pair of scissors is put into it and the other into the ectopic anus and if the bridge of tissues between them is cut. I have several times known previously incontinent children to become clean after this simple proceeding, the sphincter mechanism of the ectopic anus taking charge of the whole single opening thus produced, in the same remarkable way as happened in the case of the urethral ectopic anus in a male which is described later.

External ectopic anus in the male. A rather rare abnormality is the shifting forward of the anus on to the posterior border of the scrotum. (Figs. 8 and 9.) This must be clearly distinguished from those cases in which the " covered anus " lies in its normal position, with a sinus leading forward from it to open anteriorly ; the treatment of the two is as different as their anatomy. (Fig. 10.)

In these cases in which the anus itself is shifted forward, the principle of treatment is the same as in the analogous condition in the female ; the opening must be made to work where it lies.

Treatment. This consists in a simple backward incision and dilatation. In the case illustrated in Fig. 8, an opening had been made into the bowel in the normal anal site, and a fortunately unsuccessful closure of the ectopic anus attempted. Closure of the surgical opening and dilatation of the ectopic one gave a child, who was previously dribbling fæces continuously, an anus normal in function, situated a short distance further forward than normal. (Fig. 9.)

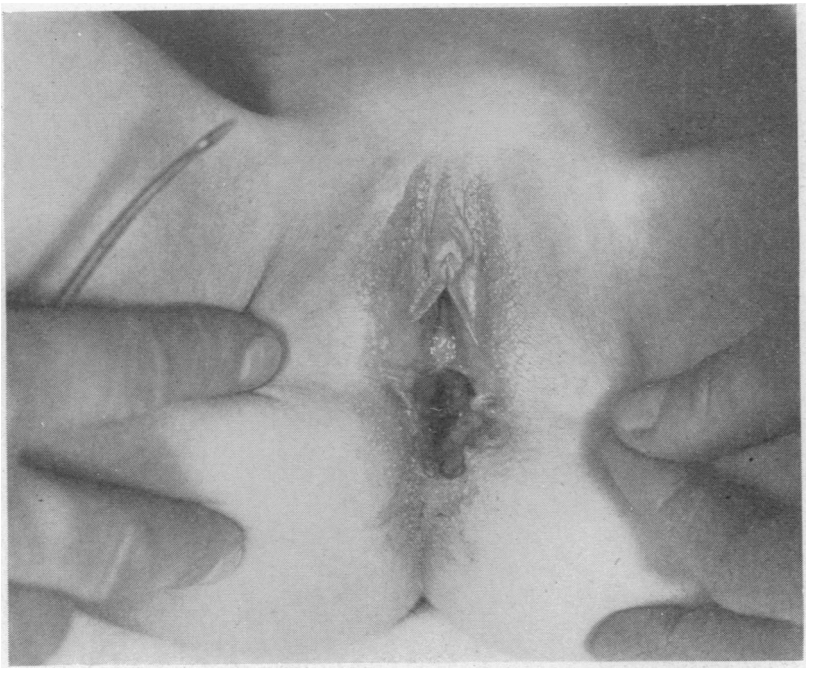

Fig. 7. " Shot-gun perineum " surgically produced by a backward incision from an ectopic anus opening low in the vagina. Control of anus is perfect.

Ectopic urethral anus in the male. I learnt a great deal from the first of the three cases of this condition which I have had to treat. All three have had associated hypospadias, with a bowel opening into the lower bulbar urethra through which gas and fluid fæces emerged freely, in contrast to the minute amounts of meconium which alone escape from the high urethral fistula. This first child was in misery from the usual fæcal fistula which had been produced by the usual incision in the anal site. I had determined after long discussion with his parents to do a controllable abdominal colostomy as an unattractive alternative to his uncontrollable perineal one. But, as a last resort, on no very clear line of reasoning, I slit up the entire perineum opening the urethra completely to the margin of the ectopic anus, and connecting this to the surgical fistula behind. To my astonishment he soon developed complete control of his motions. I thankfully left him alone for several years, far happier than he had been, but condemned to pass urine in the feminine manner. In the interval, however, I invented the method of treating hypospadias mentioned later, and finally applied it to him, with the result that the entire length of the open urethra was converted into a tube in one operation, and the child is now functionally normal.

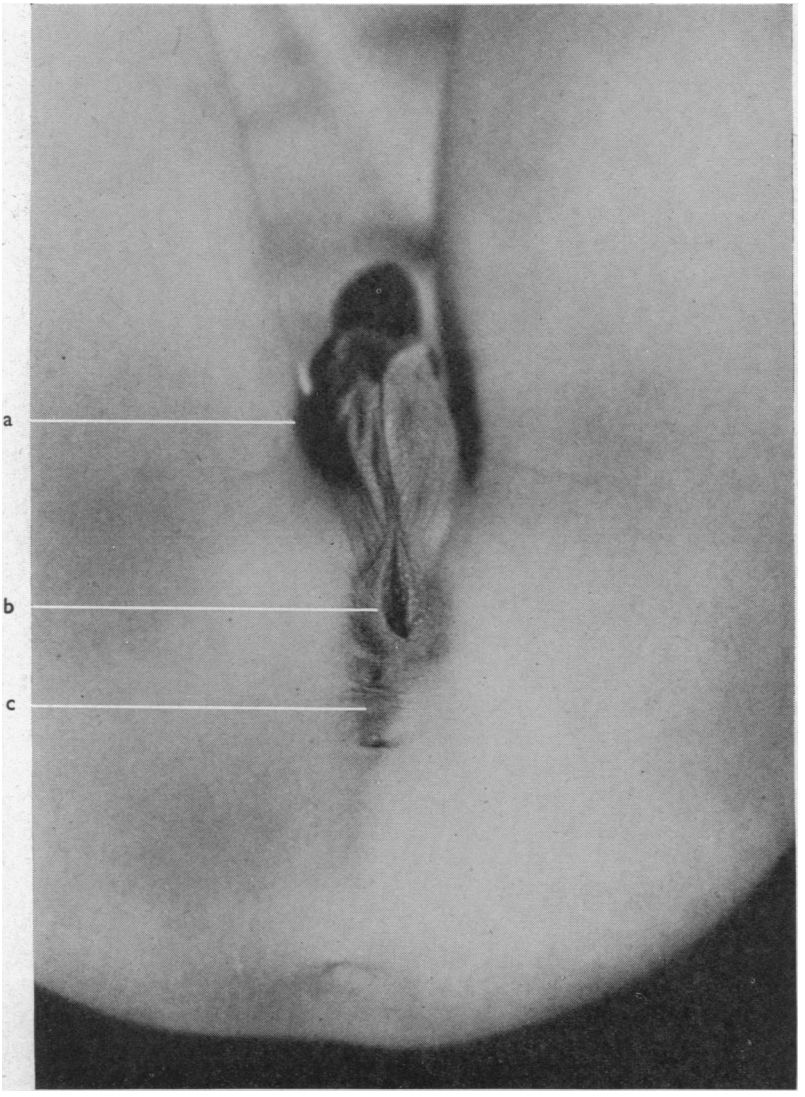

Fig. 8. Male ectopic anus. (*a*) Scrotum ; (*b*) Normally functioning ectopic anus ; (*c*) Scar of closure of surgical opening made in normal site of anus.

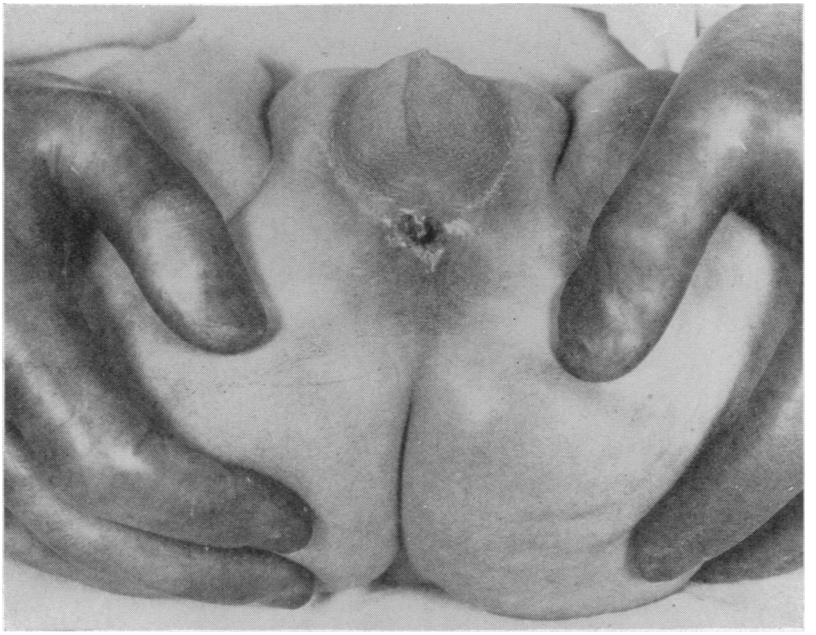

Fig. 9. Male ectopic anus dilated in its original site with excellent results. A similar case to Fig. 8, except that no attempt was made to open the bowel at the normal site.

EXCESSIVE FUSION

The covered anus in the male. This condition is described, in the few recorded cases, as imperforate anus with a scrotal or penile fistula. On inspection there is no opening on the anal site, but a narrow sinus is seen running forward from it, so superficial that the meconium with which it is filled shows as a blue line through the skin, and opening anywhere from the posterior part of the scrotum to the end of the penis.

The only explanation of this deformity, and the next one to be described, is that they are the results of an abnormal fusion of the external genital folds, extending in both directions so that the anus is covered, though normally developed, and an abnormal tube is formed running forward from it.

Treatment. This is the obvious measure of excising the sinus and exposing the anus. Here the usual surgical approach through the normal site does for once give good results, as a normal anus lies immediately below the skin. (Fig. 10.)

The covered vagina in the female. A case which is in my experience unique is illustrated in Fig. 11. There was apparently a tiny penis, with a urinary opening at its base. Posterior to this was thick skin and subcutaneous tissue, running back to the anus. At first I mistook this child for an atypical abnormal male, with a lack of development of the penis

complicating an unusual hypospadias. Mr. Douglas Stephens, however, suggested that the child was really a girl, normal except for the vagina being covered in by an excessive fusion of the external genital folds. An extensive cut backwards from what I had imagined to be the urethral opening revealed a normally formed female urethral meatus and vagina concealed underneath, and I see no reason why this child should not be functionally normal.

Adherent labia minora in the female. This very common condition is curiously little recognised and described. It consists in a filmy line of junction between the labia minora, extending forwards so as to leave merely a small opening through which urine is passed. This fusion is often so extensive that the mothers observe that " she passes water like a little boy " meaning that the stream spurts outwards and forwards owing to its impinging on this abnormal diaphragm.

It is nearly always diagnosed as congenital absence of the vagina, and I have several times known the parents to be consoled by the prospect that an artificial vagina could be constructed by " plastic surgery " when growth was complete.

Treatment. This consists in a stroke with a probe to separate the labia, followed by the gentle inunction of vaseline until the narrow raw surfaces have healed separately. If this after-treatment is omitted the condition may, of course, recur.

INCOMPLETE FUSION

Hypospadias. I have described elsewhere this condition and the operation which I have devised for its repair, so that there is no need for repetition here. It may be noted, however, that according to the classification of deformities given in this paper, there is both incomplete fusion and failure of formation in the severer degrees. That is to say there is not simply a failure of the two genital folds to fuse and form a tube, but there is also a failure of the entire underside of the penis to develop, with the result of the well known binding down of the penis into " Chordee." I know of no other deformity where a similar combination of defects consistently occurs.

Treatment. The method of treatment which I have devised, consisting in the burying of a single strip of epithelium in the knowledge that it must inevitably and spontaneously form a complete tube, has two advantages that may be mentioned. The first is that the surgeon need have no fears about opening up the urethra to any extent as a temporary measure, knowing that he will be able to close it with certainty afterwards. (See the case described of urethral ectopic anus in the male.) The second is that the knowledge that there is no need to construct an epithelial inner tube by suturing enables the surgeon to make muscular tubes intended to control the passage of fluid of a far finer calibre than is otherwise possible. The importance of this for trying to form a working sphincter to the bladder is obvious.

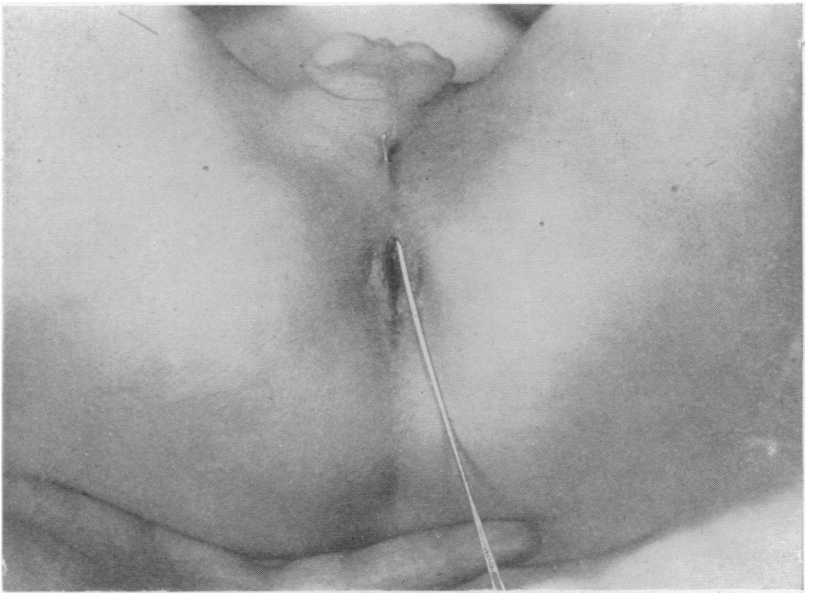

Fig. 10. Covered anus in male. In this case originally the only opening was where the tip of the probe emerges. A surgical opening was made, and a normal anus discovered under the skin of the normal site. Subsequently the sinus was excised and only a close inspection reveals any abnormality at all.

Congenital enlargement of the urethra in the female. A continuous series of deformities in the female can be found, ranging from a slight increase in the diameter of the urethra to a complete extroversion of the bladder. I am sure that the collection of series of cases of gradually increasing severity is a method of great importance in the study of many deformities. (*Cf.* Hare-lip or talipes.) The only case of this kind which I have seen was that of a girl suffering from complete incontinence, who was found to have a urethra of a size that would readily admit an ordinary lead pencil.

Treatment. The crude device of tying a catgut suture round the urethra, in which a urethral catheter was kept for a week to drain the bladder, gave complete control.

Pubo-vesical cleft in the female. I have had three cases of small girls in whom there was an opening into the bladder which would admit two fingers readily. The condition is obviously, it seems to me, a mild degree of extroversion of the bladder, but as the bladder is not extroverted the name can hardly be used. (compare the condition of " talipes equino-varus," the most severe degrees of which have the foot in calcaneus.) Stiles has called it " epispadias in the female," a name which also has obvious disadvantages. As the apparent failure of fusion involves both the pubis and the bladder, I suggest the name of " pubo-vesical cleft."

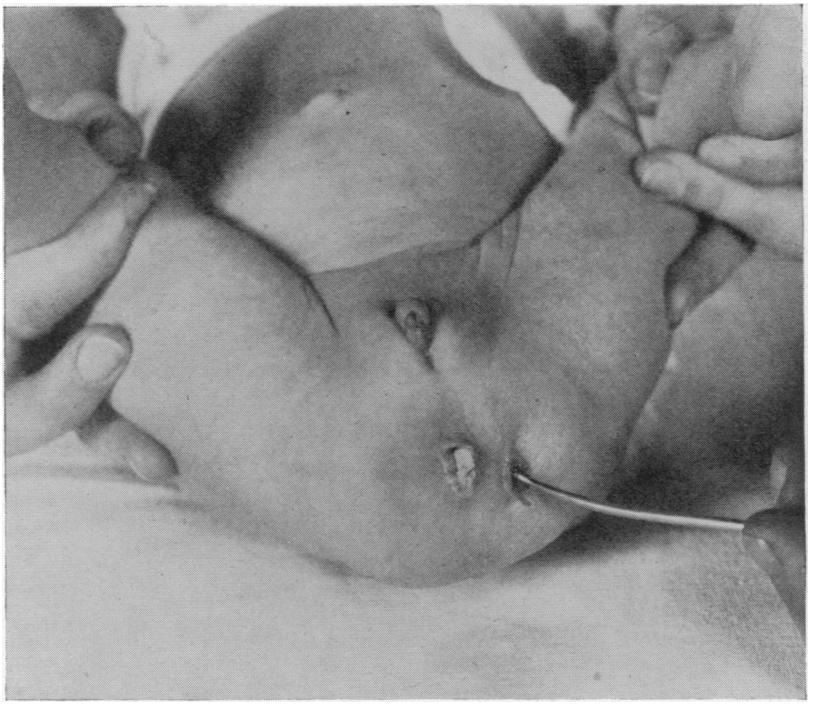

Fig. 11. Covered vagina. The probe points to an abscess which had no importance to the deformity. Incision of the band of tissue between the clitoris and the anus revealed a normal vagina.

Treatment. In all three of these I decided to work on the hypothesis that the normal constituents of the bladder and its sphincter mechanism were present. Accordingly, I freed both sides of the cleft in the mons veneris, and then dissected out the wall of the bladder round where the neck and urethra would have lain normally. The bladder muscle was then sutured up tightly round a minute strip of mucosa, all the rest of the bladder lining in the neighbourhood having been removed. The raw edges of the cleft mons were then brought together by " double-stop " sutures and the wound left without drainage of the bladder. The result in two of the three was complete control of micturition, after a period of stress incontinence. In the third, a nervous child of three, I intend to operate again if no signs of control appear within a year or so.

Penile epispadias in the male. This condition is inexplicable on any of the current embryological hypotheses, and I hope to discuss the difficulties elsewhere.

Treatment. The technique of suturing the outer integuments only, leaving a strip of mucosa deeply covered at the bottom of the opposed raw surfaces, answers as well in this condition as in hypospadias.

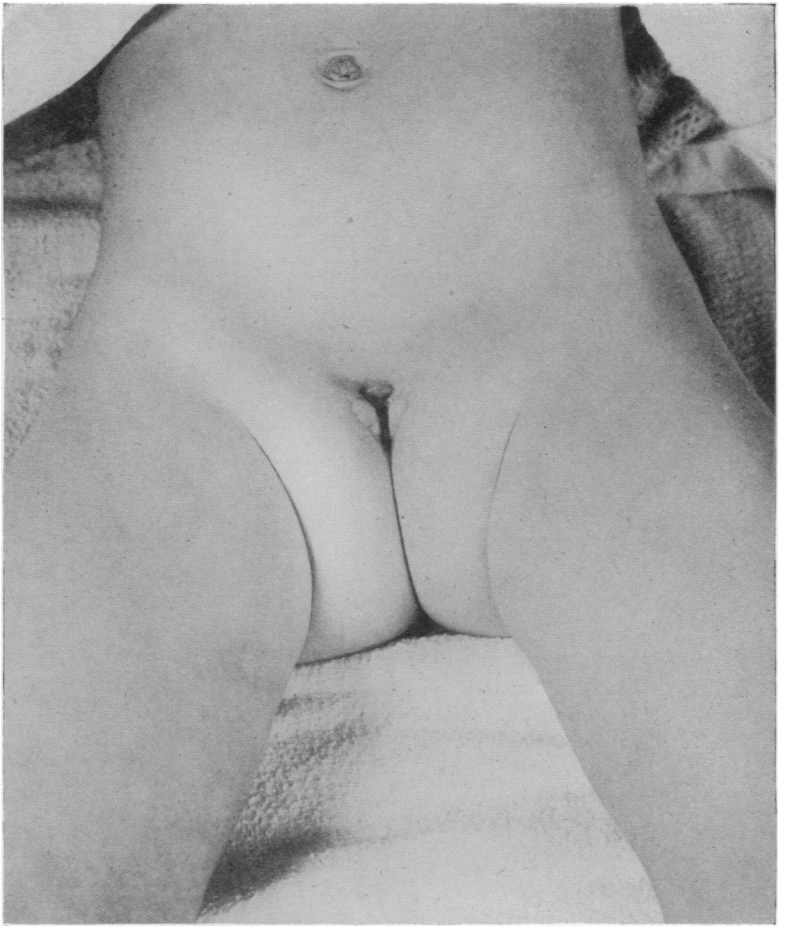

Fig. 12. Pubo-vesical cleft in female. The mons veneris is cleft and one can look directly into the bladder with a speculum.

Perineal drainage is, of course, impossible and I am not sure that drainage of the bladder in these cases is necessary. However, I usually keep in a urethral catheter for a week or so.

Penile epispadias with pubo-vesical cleft in the male. This condition is usually described as epispadias with incontinence, but I prefer a term that relates it to the similar condition found in the female. The penile part of the deformity is the same as in the simple epispadias, but above this there is an opening into the bladder that is always greater than normal, and that may be big enough in a half-grown boy to admit an adult finger with ease. Urine, of course, dribbles continuously, and most cases end up either with highly unsatisfactory urinals or with ureteric transplants into the bowel.

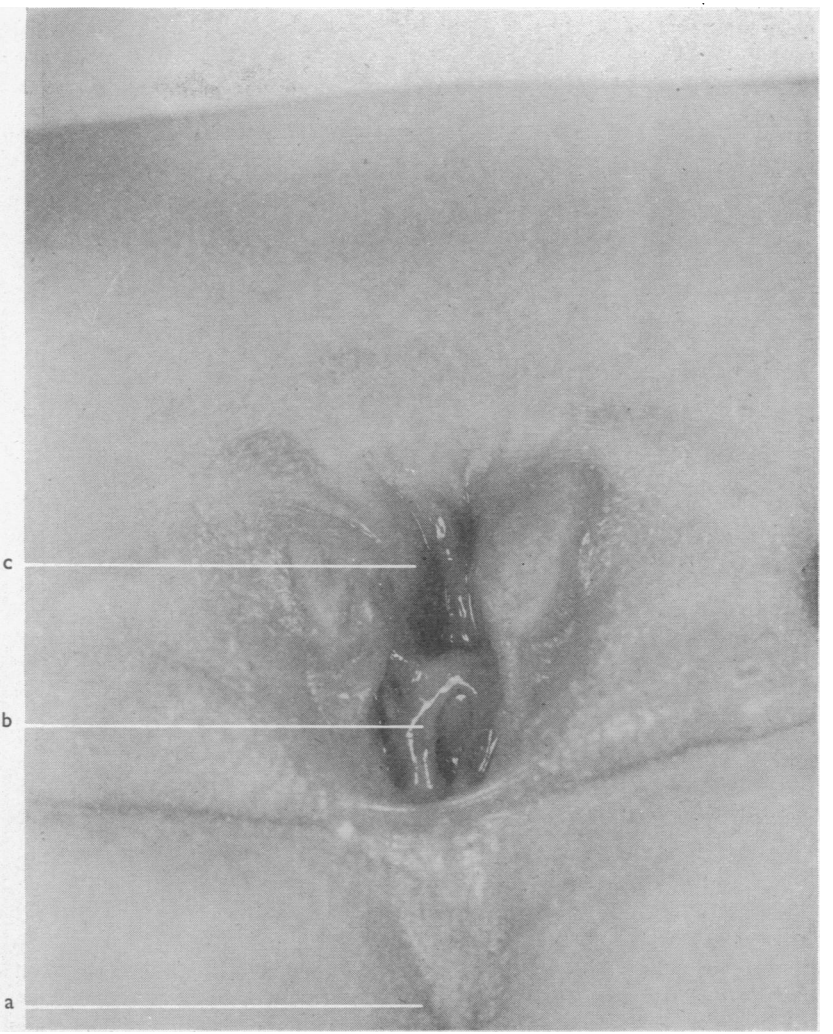

Fig. 13. Pubo-vesical cleft. Close view of case similar to Fig. 12. (*a*) Anus; (*b*) Vagina ; (*c*) Opening into bladder.

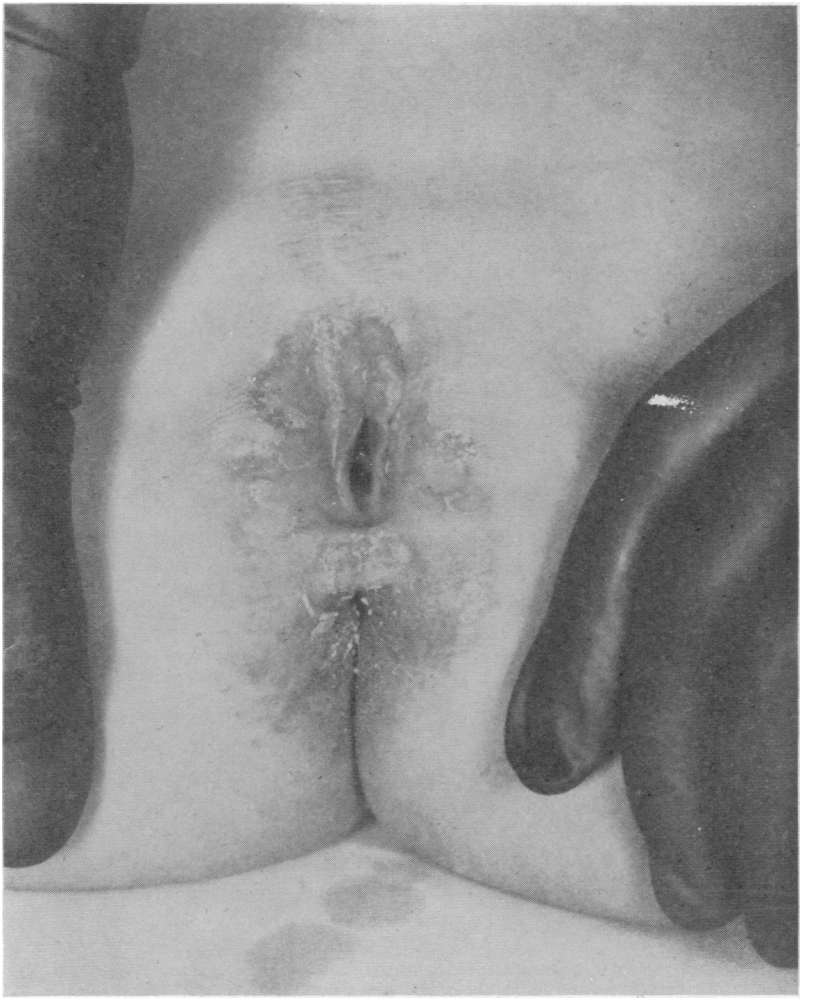

Fig. 14. Case shown in Fig. 13 after closure of cleft. The child now has complete control of urine.

Treatment. It is almost invariable, in my experience, for the easier part of the problem, the construction of a penile urethra, to be attempted first. Whether this is successful or not, and it is usually not, it very much hinders any subsequent attempt at the relief of the patient's main trouble of incontinence, by scarring up the huge urethra, which when untouched allows inspection of the site where control of urine should be exerted.

The site of control of the bladder lies in the male in a most inaccessible region, and one that has in addition no easy lines of dissection and a profuse and intractable blood supply. I have attacked this problem with success in two cases, gaining access to the neck of the bladder by dividing the pubic symphysis and subsequently suturing it by thick chromic catgut. The neck of the bladder when exposed was treated as in the female cases of pubo-vesical cleft which I have mentioned, excising all the mucous lining with the exception of a very narrow strip, and suturing the muscle tightly round a ureteric catheter. Suction drainage of the weight of two feet of water was kept on both the bladder and the depths of the wound, which otherwise would become sodden with urine.

In both cases control was slow to develop, taking about a year, and was probably much helped by contraction of the scar tissue around the neck of the bladder.

I have tried to narrow the sphincter from inside the bladder as described by Young, but without success; and I regard the external approach which I have described as giving far better chances of the really drastic surgery which I consider necessary.

Extroversion of the bladder in both sexes. This dreadful condition is, as has been pointed out, the last and most severe of a series of deformities starting with congenital enlargement of the urethra in the female, and with glandular hypospadias in the male.

Treatment. I have not given up trying to get a success by reconstructing the bladder and sphincter. It is certainly possible to sew the bladder into a sac with a narrow neck, and get a skin covering for this. In this way, even in the extremely likely event of complete failure, one of the most difficult problems in the surgery of this condition will have been solved. This is the treatment of the cleft bladder itself after the urine, as is now our only reliable resource, has been diverted into the bowel.

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